ABSTRACT OF THE INVENTION

The present invention relates to identification of a gene that is inactivated in a mucolipidosis condition. In particular, the invention concerns mutations that disrupt a mucolipin, preferably MCOLN1, in mucolipidosis IV. Recombinant nucleic acids encoding mutant forms of MCOLN1, oligonucleotides specific for such mutations, and diagnostic and therapeutic applications related to these discoveries, are also contemplated.